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A NEW FISH ASSAY TO DETECT STRUCTURAL AND NUMERICAL CHROMOSOME ABERRATIONS IN HUMAN SPERM; DATA FROM HEALTHY MEN AND A TRANSLOCATION CARRIER.

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Both structural and numerical chromosome aberrations in sperm represent important categories of paternally transmitted genetic damage. Therefore, a new multi-probe FISH method, using four DNA probe on three targets, was designed to detect three types of chromosomal defects simultaneously: sperm carrying terminal duplications or deletions in chromosome 1p, diploid sperm, and aneuploid sperm involving chromosomes 1 or 8.

Baseline levels were determined for 4 healthy donors. Among 150,354 sperm analyzed, the average frequencies of telomeric duplications and deletions of 1p were 3.7 ± 2.3 per 10^4 sperm (range: 1-9) and 2.5 ± 3.3 (range: 0-13) respectively. Average frequencies of disomic sperm for chromosomes 1 or 8 were 1.9 ± 2.4 per 10^4 (range: 0-8) and 7.1 ± 4.0 for diploid sperm (rang: 1-14). Reproducibility between hybridizations and multiple samples from the same donor was demonstrated. The different kinds of chromosomally abnormal sperm from a t(1;10)(p22.1;q22.3) translocation heterozygote were analyzed to further evaluate our new FISH assay. All 2:2 segregation types and one 3:1 type were detected. The frequencies observed using FISH were in agreement with earlier data reported using the hamster-egg technique. These findings indicate that the proposed FISH assay can be used to detect structural and numerical chromosome aberrations in human sperm in a variety of genetic, physiological and toxicological applications.

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